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Early identification of birth defects can reduce secondary disabilities in newborn infants

Birth defects and developmental disabilities are important issues of public health concern.[1] Birth defects are abnormalities of structure, function, or metabolism that are present at birth and can result in physical or mental disability, or death.[2–5] About 3% of children born in the United States (US) have a major birth defect; these account for about 20% of all infant deaths.[4] Disability is defined as a limitation of activity associated with long-term physical, sensory, and/or cognitive impairments.[1,6] Developmental disabilities can begin *in utero* or after birth because of injury, infection, or other factors.[5] About 17% of children in the US have a developmental disability, with about 2% having a disability severe enough to require life-long care and special services.[1]

In 1992, the UN General Assembly proclaimed December 3rd as the International Day of Persons with Disabilities.[7–9] This day is celebrated every year to promote awareness and understanding of the problems faced by people with disabilities all over the world.[9] The goal is to promote inclusion, dignity, and the rights of affected people all over the world, be it in political, social, economic, and cultural life.[10] We need to be "United in action to rescue and achieve the Sustainable Development Goals (SDGs) for, with, and by persons with disabilities."[11,12] Unfortunately, much more work is still needed to achieve the SDGs by the original timeline of 2030.[13] The Children's Health Act of 2000 recognized the relevance of these conditions with the creation of the National Center on Birth Defects and Developmental Disabilities (NCBDDD) at the Centers for Disease Control and Prevention (CDC).[14,15] It has recently launched a national campaign, "Learn the Signs. Act Early," to educate parents and healthcare providers about the importance of early, timely intervention.[16–19]

We need a fundamental shift in our commitment, solidarity, and financing to reduce the public health burden emanating from these issues.[20] A political declaration at a recent SDG Summit[21] focused on the achievement of sustainable development and shared prosperity for all, by defining policies and actions that target the poorest and most vulnerable, including persons with disabilities.[22] The UN Disability Inclusion Strategy (UNSID; June 2019)[23] aims to raise the Organization's standards and performance on disability inclusion.[24,25] The idea was to provide sustainable and transformative progress in at-risk populations.[26]

Our journal, the *newborn* aims to cover fetal/neonatal problems that begin during pregnancy or occur after birth during the first 1000 days after birth. In this 4th issue of the second volume, we present 8 new articles (**Figure 1**). In neonates, Down syndrome is the most common genetic cause of intellectual disability; it affects approximately 1 in every 700 children, and accounts for around 15–20% of the intellectually disabled population.[27] The condition was first described by an English doctor, John Langdon Down,[28] and was subsequently associated with the trisomy of chromosome 21 by Professor Jerome Lejeune, a geneticist in Paris.[29,30] Since then, other less frequently seen forms of the condition have been discovered. Approximately, 94% of people with Down syndrome have standard trisomy 21, but 4% have a translocation and 2% have a mosaic Down syndrome.[31] In most cases, Down syndrome is not hereditary; it affects people of all ethnicities, religious backgrounds and economic situations.[32] It is covered under the Social Security Administration (SSA)'s "Blue Book" of impairments under Section 110.00.[33] In this issue, we bring a short communication from Dr Amita Garg, who is a renowned cardiologist in New Delhi, India, and is also a mother who has raised a child with trisomy 21.[34] She has first briefly shared her personal experience and then her views on how we medical professionals could/should interact with families in a positive and encouraging way. She has also listed resources that can help parents who are raising a child with this condition.

In another article, Singh *et al*.[35] have described the pathogenesis and implications of sensorineural hearing loss in children due to congenital CMV infections. These *in utero* infections account for nearly 25% of childhood hearing loss by the age of 4 years.[36] Hearing loss during childhood is an important disability with secondary effects on speech development and acquisition of linguistic skills.[37] A

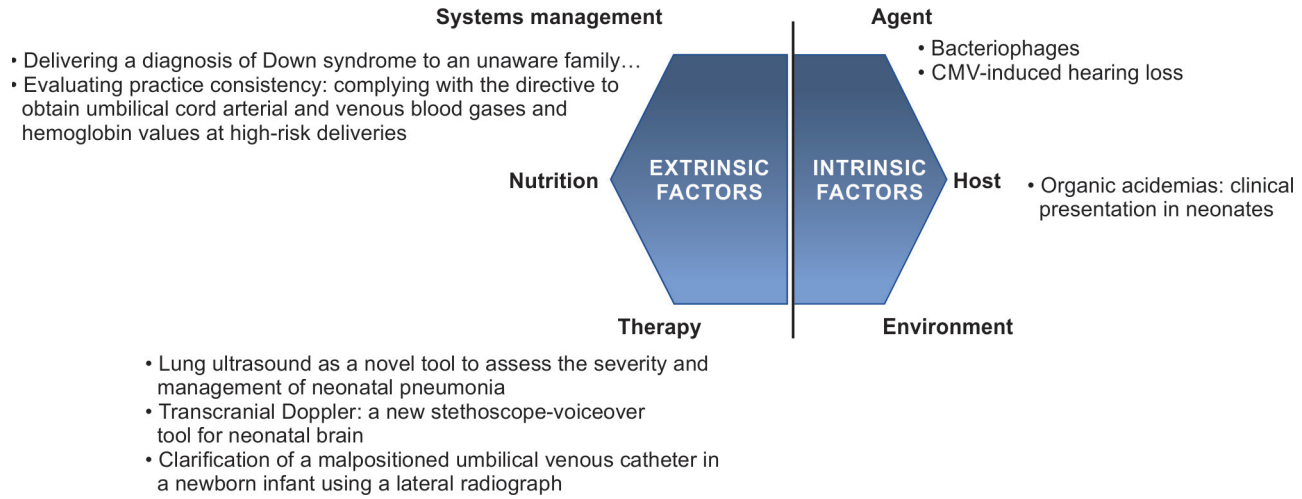

Fig. 1: **Areas of focus in the *newborn*, Volume 2, Issue 4.** We have expanded the traditional agent-host-environment trinodal disease model to a hexagonal system. The three additional foci represent extrinsic factors that can affect health—those originating in therapy, nutrition, and systems management. This issue covers 4 of these foci, namely infectious diseases, host factors, treatment/monitoring systems, and systems management.

multidisciplinary approach is required; many children may need assistive hearing devices or cochlear implantation depending on the severity of their hearing loss.[38,39] In addition, early intervention services such as speech or occupational therapy may be needed.[38]

Kaushal and coworkers[40] have contributed with a detailed review of organic acidemias (OAs), which are heritable genomic abnormalities that result in accumulation of toxic organic acids. Most patients with severe deficiencies in the involved enzymatic pathways can become symptomatic in early infancy.[41,42] Acute clinical features include liver failure, lethargy, altered sensorium (encephalopathy), and/or seizures in the acute phase; subacute/delayed manifestations may include failure to thrive, developmental delay, and/or cardiomyopathy.[41,43,44] These manifestations can resemble those seen in systemic inflammatory response syndrome related to sepsis and a high index of suspicion is needed for timely diagnosis.[42]

In an interesting article, Tweddell and colleagues[45] report their experience with efforts to comply with the directive to obtain umbilical cord arterial and venous blood gases, and hemoglobin values at high-risk deliveries and then evaluate practice consistency.[46] They analyzed data from 1,050 births with placental abruption from a 24-month period. About 70% had both a cord arterial and venous gas, and hemoglobin levels reported. Acidosis was noted in 14%. In this subset, nearly 80% had abruption confirmed after birth. Fetal/neonatal anemia[47,48] was diagnosed in 12%. There is an opportunity to improve compliance with the directives to obtain cord arterial and venous blood gas and hemoglobin at high-risk births;[49] this may allow rapid evaluation of about 30% more high-risk infants for the presence of acidosis and anemia at birth.

Two articles emphasize the increasing importance of bedside sonography for monitoring critically ill infants. In the first, Kumar and Patodia[50] have reviewed cerebral Doppler ultrasound for tracking cerebral perfusion for monitoring critically ill very premature infants. They performed a comprehensive literature search using two databases and have provided an overview of several cerebral Doppler parameters such as the resistive index in the anterior cerebral artery in monitoring the evolution of neonatal disorders and/or response to therapy.[51–53] These data can also possibly help in predicting long-term neurodevelopment outcomes.[54] In another article, the same group has reviewed the importance of lung ultrasound as a tool for monitoring the severity of pneumonia in critically ill neonates in neonatal intensive care units. Durga *et al*.[55] have proposed a lung ultrasound (LUS) scoring system to help monitor the severity of pneumonia and its progression. Such bedside scores have been useful in adults with COVID-19 disease[56–58] and could help monitor the changing severity of lung disease in infants.

Singh *et al*.[59] have contributed another article focused on bacteriophages, viruses that invade bacterial cells.[60,61] Phages show a remarkable degree of diversity; recent advances in viral metagenomics show an unprecedented catalogue of phages in all microenvironments.[62] Phages may contain double-stranded (DS) DNA, single-stranded (SS) DNA, SS-RNA, and DS-RNA.[63] There is also a very high degree of structural diversity.[64] In terms of biological targets, these viruses attach and kill specific bacteria by expressing endolysins and holins without affecting the commensal microflora.[65,66] At the same time, bacteria are also developing numerous defense mechanisms to inhibit the phage life cycle.[67] Phages can inhibit some of these bacterial defenses, and the battle could go on.[68] These viruses may have translational importance with phage-based treatments; single phages, phage cocktails, phage-derived enzymes, phages in combination with antibiotics, and genetically modified phages might be useful in treating bacterial sepsis, even multidrug resistant (MDR) pathogens.[69,70]

Finally, Bottu and colleagues[71] submitted an interesting set of radiographs from a newborn infant in whom an umbilical arterial and an umbilical venous catheter (UVC) were inserted for stable vascular access. An anteroposterior radiograph showed the UVC was coiled up in the liver. Several possibilities including vascular abnormalities came to mind, but a lateral radiograph removed these doubts and was reassuring. The catheter was promptly removed, and the subsequent hospital stay of the infant was uneventful. They have reported these findings to re-emphasize that lateral radiographs can be useful.[72]

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