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EDITORIAL

Inborn Metabolic Disorders: The Winding Path Ahead, in the Road Less Traveled

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Inborn metabolic disorders (IMD), previously known as inborn errors of metabolism (IEM), though individually rare, collectively contribute to incidence of at least 40 per 100,000 live births [1]. In a country like India, it contributes tremendously to morbidity and mortality, leading the great Prof. IC Verma to refer to it as a "burden". He emphasized an accelerated shift to non-communicable diseases, like congenital abnormalities and genetic disorders even in India in this article published at the turn of this millennium [2].

Back in 1950, communicable diseases seemingly contributed the major share to infant mortality rate (IMR) in India. The infant mortality rate has reduced from ~189 per 1000 live births in 1950 to ~27 per live births in 2022, largely attributable to the improvement in healthcare facilities, control of communicable diseases as well as improvement in management of certain non-communicable diseases, like congenital heart diseases (CHD). This was possible through a concerted effort from doctors (pediatricians, neonatologists and cardiologists) government agencies, celebrities, newspapers and various other sections of the populations who have spread the awareness about communicable and non-communicable diseases like CHD, which might have been unimaginable 50 years ago [3–6].

A similar concerted effort is needed to bring IMDs under control. Progress has been achieved to different extents, thanks to various government and non-government initiatives like ICMR task forces and multi-centric projects, DBT Nidan Kendras, rare diseases groups, and even individuals who have experienced the impact of these disorders, directly or indirectly. However, even after 2–3 decades, in India we're still in the infancy with regard to implementation of newborn screening (NBS) or spreading public awareness about IMD. Even pediatricians/neonatologists stand divided in their attitude. The public perception of IMDs has to change drastically. Biochemists have an important role to play in this regard, all biochemistry postgraduates can contribute by educating the young I MBBS students or help conduct NBS across populations.

The challenge is regarding the cost of implementing state-of-the-art technology. tandem mass spectrometry (TMS) is currently available only to a limited number of laboratories. In addition to the cost, training of personnel poses an additional challenge. With a more concerted approach towards IMD, involving the corporate providers and innovative solutions towards nation-wide implementation of NBS, changes are just over the horizon.

International Federation of Clinical Chemists (IFCC) along with the International Society for Newborn Screening (ISNS) has formed a task force for newborn screening (TF-NBS) to look into the feasibility of carrying out NBS globally, across various developing countries, including India. TF-NBS is currently looking for ways of implementing screening of, at least, common IMDs globally. This task force has formed clear mandates and a vision for moving ahead. [7]

In India, this is feasible using techniques like DELFIA, provided there are at least a few referral laboratory in the country where follow-up testing could be carried out. The projects, though ambitious, are definitely feasible; however it requires tremendous amount of coordination, and, as mentioned before, concerted effort from all stakeholders. This is already being carried out in India, as mentioned before, to different extents, in a few centers. However, a nation-wide

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NBS program, even for a single disorder, seems a distant dream.

The criticisms against giving additional health priorities for IMD have always been there; they're untreatable, the children are a 'burden' to the families and societies, it's impossible to counsel the families, pediatricians/neonatologists have to time when they're already overburdened with many other 'more important' problems, and for the providing companies, there's 'no business interests'. However, most of these are just myths, and we, biochemists, can help bust these myths. We can help mobilize influential segments of the public, celebrities, and/or government agencies and help educate the masses through them, newspapers and through our own lectures, writings etc.

Almost all biochemists have a career in IMDs as their dream, but many are unsure about the path ahead. Individuals with IMDs may not even know how to ask for help. Or, for that matter, they might never realize whether they're truly a burden, to their near and dear ones, or to their societies! If we can contribute even that little bit to helping one individual with IMD in our career, we should try to do that. Needless to say, early diagnosis is one of the important ways by which we can help save an IMD patient, and that is why the role of a biochemist is vital.

The path ahead is definitely long and tortuous, but with a few individuals who can work in a determined manner, there's always, as they say, light at the end of the tunnel. It's a road less traveled; who knows what you might discover. If you don't take the path, you never know that!

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