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Outline of New Born Baby Checking of Neurological Illnesses

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Abstract

More than 400 different disorders fall under the umbrella of neuromuscular diseases, which are diverse in their phenotypic range. Due to their rarity and complexity, neuromuscular illnesses are frequently only discovered after a very long period of time, at which point irreparable muscle loss may reduce the effectiveness of treatments, when any are available. Neonatal screening may offer an answer for early detection and therapy in this situation. PRISMA standards were followed to perform a systematic evaluation of the literature in PubMed up to May 1, 2021, which included conditions with a clear involvement of the peripheral nervous system and classic neuromuscular illnesses (including central nervous system disease with severe neuropathy). We identified seven disorders for which newborn screening data were available: Krabbe disease, X-linked adrenoleukodystrophy, Pompe disease, Duchenne muscular dystrophy, spinal muscular atrophy, Myotonic dystrophy type, Krabbe disease, X-linked adrenoleuko- dystrophy, Pompe disease and metachromatic leukodystrophy. Changing from a biochemical to a genetic-based approach will require a global technical shift in newborn screening for neuromuscular diseases. In order for innovative medicines to be as effective as possible, the rapid development of therapy also necessitates the ability to quickly adjust the list of ailments treated.

Keywords: Duchenne muscular dystrophy; Pompe disease; Neuromuscular problem; Infant screening

Introduction

A stricto sensu mid-career 45-year-old mycologist developed the idea of screening neonates for neuromuscular disorders and advocated Creatine Kinase (CK) dose for Duchenne muscular dystrophy (DMD) forty-five years ago. Newborn screening (NBS) for phenylketonuria had only recently been introduced in the majority of affluent nations at the time. This essay is a celebration of the 90th birthday of this former mid-career myologist. 45 years later, the Dubowitz disease (not to be confused with Dubowitz syndrome), also incorrectly referred to as "spinal muscular atrophy type 2" by a very small number of physicians, has established itself as the neuromuscular fields ideal example of the ideal indication for NBS [1].

More than 400 different disorders fall under the umbrella of neuromuscular diseases, which are diverse in their phenotypic range. For the majority of them, there were few disease-modifying therapies accessible until recently. However, with a better understanding of pathophysiology and preclinical research, some transformative medicines have had a significant impact on both inflammatory disorders and hereditary diseases as Brown-Vialetto-Van Laere syndrome, spinal muscular atrophy, and congenital myasthenia (BVVL) [2]. Additionally, promising early results have been reported for DMD, for which five medicines have so far received regulatory approval, X-linked myotubular myopathy, and limb girdle muscular dystrophy. Due to their rarity and complexity, neuromuscular illnesses are frequently discovered relatively late during which time irreparable muscle damage may restrict the dramatic effectiveness of early treatment given to patients. Before a proper diagnosis is made and the right course of therapy is suggested, the protracted diagnostic procedure can result in decades of quality of life impairment even in the absence of a muscle degradation process, such as in some forms of CMS. The modified Wilson and Jungner criteria, which are frequently used to assess whether screening for a condition should be included in an NBS panel, serve as the general guidelines for newborn screening throughout the world. These ten objects are included in this list. The sought-after ailment needs to be a serious medical issue. Patients with known diseases should receive a recognized treatment. There should be resources for diagnosis and treatment [3]. A discernible latent or early symptomatic stage should exist. A proper test or examination should be available. The general public should accept the exam. It is important to have a thorough understanding of the condition's natural history, particularly how the disease progressed from latent to diagnose. On whom to treat as patients, there should be consensus.

In comparison to potential spending on medical care as a whole, the cost of case-finding (including patient diagnosis and treatment) should be economically balanced. Case-finding need to be an ongoing procedure rather than a one-time undertaking. In comparison to the US, the European Union (EU) and the United Kingdom take a far more cautious stance when applying these criteria [4]. As a result, there are major differences in the types of diseases that are screened across nations or even within the same nation. According to these standards, in 1976 our mid-career myologist noted that "the stage does not yet seem set for a [UK] nationwide programme of screening for preclinical Duchenne muscular dystrophy, but when the time is ripe for it the techniques will hopefully be sufficiently standardised for immediate application [5]." NBS has been set up for the past 60 years as a metabolic and endocrine screening technique, however many hereditary neuromuscular illnesses that are curable in children, including BVVL, CMS, or SMA, lack metabolic or endocrine markers, adding to the difficulties in screening. However, the stark contrast between pre-symptomatic and post-symptomatic treated SMA patients, the widespread success of NBS for SMA, and the pipeline of potential therapies all imply that NBS may be effective against a number of neuromuscular diseases before our previous mid-career myologist celebrates his 100th birthday. In this context, we reviewed the official or pilot NBS initiatives that are

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now running in the field of neuromuscular illness [6].

Methods

Literature Search

Using the PRISMA checklist, a literature search was carried out using Medline (PubMed) [7]. After Prof. Victor Dubowitz turned 70, we looked for original, full-text articles reporting NBS programme in neuromuscular disease (Aug 06th 2001). Neonatal screening, dried blood spot testing, dried blood, and Guthrie are a few examples of key terms associated with NBS that were combined with key terms for neuromuscular illness to find relevant articles. In Supplementary file 1, the detailed search plan is schematically illustrated. Until May 1 2021, a literature search was carried out [8].

Selection of Studies

Titles and abstracts were independently reviewed for eligibility by two researchers (TD, LS), who then assessed the complete text [9]. The publications had to be original research that had been published in either English or French, and they had to describe an NBS programme for at least one neuromuscular disorder or a condition that clearly included the peripheral nervous system (mostly peripheral neuropathy). Following a comparison of the two reviewers' findings, a list of studies for full-text screening was produced. The reasons for each article's omission were noted, and any potential conflicts were identified for consensus resolution [10].

Results

Study selection process

405 papers describing NBS for neuromuscular illnesses were found in the initial searches. 84 papers were selected for full-text screening after deleting 108 duplicates and screening by title and abstract [11]. 36 full-text studies were validated as eligible, and 8 articles identified by bibliography were added. The process used to identify these studies is shown in Supplementary File 2 and is based on the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) recommendations [12].

At times, you might not notice neurological symptoms until your child begins missing milestones [13]. Here are some examples of developmental milestones your child could miss:

1) The ability to hold his or her own head up

2) The ability to roll over, sit up, crawl, or learn to walk

3) Learning to feed him or her

Sometimes you might have difficulty putting your finger on the problem, but you simply know that something is not right with your child. It's important to seek medical help if your child is experiencing any of these problems. Fragile and premature newborns can get MRI-compatible incubators at UPMC Children's Hospital, unlike at many other centers [14]. This means they can get expedited testing without having to worry about compromising their body temperature. This is very important for their overall well-being [15].

Advanced fetal brain magnetic resonance imaging is available for detection of neurological conditions even before the baby is born. This allows our team to meticulously plan your infant's birth so that efficient treatment is instituted immediately after delivery. These tools also help in early identification of neurological disorders that may not be apparent at birth and can present later [16].

Discussion

We identified seven disorders that have been the focus of NBS over the past 20 years and clearly involve the peripheral nervous system. Given the significance of early intervention proposed or demonstrated in all clinical developments and the striking effectiveness of pre-symptomatic therapy SMA is undoubtedly the disease for which NBS has the broadest consensus [17]. Interestingly, SMA NBS programmes were started before disease-modifying drugs were approved, but they have helped to show how effective early treatment is. The NBS programme is very cost-effective, even though this has yet to be explicitly proven, given the low cost of screening and the very large societal cost of untreated sickness or post-symptomatic disorders. The treatment protocol must yet be defined, taking into account the challenging issue of patients who exhibit symptoms at birth and, on the other hand, the spectrum patients who have four copies of SMN2 [18].

Conclusion

Despite a new agreement favouring the early treatment of patients with four copies, there is currently no unambiguous and widespread opinion regarding the management of these patients. The most important technical and prognostic factor that NBS of Pompe disease brings is the fraction of late onset forms at birth, for which there is now no evidence of early therapy, as opposed to the infantile form, for which a treatment should unquestionably be started. In contrast to SMA, where the more severe form makes up roughly 60% of all forms, a recent study from Pennsylvania has shown that the earlier form is less common than the later forms in a ratio of 1:15. Some neuromuscular illnesses, as outlined in the present review, are currently amenable to NBS because they can be distinguished by a sensitive biochemical assay or by a particular hotspot mutation (e.g., deletion of SMN1 exon 7 in SMA). However, neither a distinctive biomarker nor a common molecular flaw is present in the majority of NMDs. Therefore, the biochemically based NBS platforms currently in use are not appropriate for screening for these illnesses. There are two distinct examples in CMS and BVVL. 34 genes have been identified as CMS contributors thus far.

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Conflict of Interest

No potential conflict of interest relevant to this article were reported.

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