

## Short commentary on False Positive New-Born Screening Results

**Andrea Mazzanti**

Scientific Clinical Institutes of Maugeri IRCCS, Pavia (Maugeri Foundation), Italy

### Short Commentary

Journal of Child and Adolescent Health commemorates its 3 years-service to the scientific community by consistently publishing peer-reviewed articles and tracking the progress and significant studies related to child and adult health. Ever since its inception, releasing its regular issue on a half-yearly basis, this transdisciplinary journal is also releases special issues and conference proceedings from time to time, thus comprehensively covering paediatrics and its sub-specialties, child healthcare policy and practice, neonatal health, child immunization and immunology, nutrition, early detection of diseases in child and adolescents, mental developments, genetical problems, psychology and mental disorders associated with child and adolescents, social and family life of child and adolescents, youth development, epidemiology, substance abuse, physical disabilities, bullying, infant and young children medication, psychotropic medication and psychotherapy, and other health problems and treatments related to child and adolescents. The journal strives to publish the original research articles, epidemiological studies, new methodological clinical approaches, case reports, design and goals of clinical trials, review articles, points of view, editorial's, and Images. In this editorial one of the recent and impactful research articles on neonatal screening false positive results which is published in our journal will be discussed.

Newborn screening (NBS) programs for treatable disease have long been established in an effort to reduce associated morbidity and mortality with early identification and intervention. Bob Guthrie championed the NBS program through his innovative approach to testing for Phenylketonuria (PKU) using dried blood spots on filter paper [1]. By 1965, 32 states had enacted laws to screen for PKU using Guthrie's methods [2]. Recognizing the widespread acceptance of Guthrie's screening methods, James Maxwell, Grover Wilson and Gunner Jungner identified classic screening criteria in their 1968 World Health Organization bulletin [3].

NBS programs were further revolutionized with the introduction of tandem mass spectrometry (MS/MS), which allowed for multiple diseases to be tested [4]. In 2006, the American College of Medical Genetics recommended that expanded newborn screening include 29 inherited diseases [5]. Authors from their research statistical approach stated that limitations to their study included the inability to assess the true

false negative rate, therefore the assumption of 100% sensitivity, as a consequence of the inability to obtain long-term follow-up information on all patients admitted to the NICU. In addition, the broad categorizations used in the study design (e.g., AA and AC) limited the ability to identify particular patterns of analyte abnormalities and their associated diseases (e.g., homocystinuria and methylmalonic academia), data that could be critical to inform future interventions. Perhaps most importantly, our study was limited in scope, focused on practice in a single unit in a single state. Despite these limitations, the data presented suggests a significant problem that could have serious consequences, and it is our hope that these findings will foster a national conversation focused on improving NBS in the NICU population.

### References

1. Levy HL. Historical perspectives: Newborn metabolic screening. *Neo Reviews*. 2005;6(2):57-60.
2. Guthrie R, Susi A. A simple phenylalanine method for detecting phenylketonuria in large populations of newborn infants. *Pediatrics*. 1963;32(3):338-43.
3. Wilson JM, Jungner G. Principles and practice of screening for disease. World Health Organization, Geneva, Switzerland. 1968.
4. Naylor EW, Chace DH. Automated tandem mass spectrometry for mass newborn screening for disorders in fatty acid, organic acid, and amino acid metabolism. *J Child Neurol*. 1999;14(1):4-8.
5. Watson MS, Mann MY, Lloyd MA, et al. Newborn screening: Toward a uniform screening panel and system-executive summary. *Pediatrics*. 2006;117(3):296-307.

### Correspondence to:

Andrea Mazzanti

Scientific Clinical Institutes Maugeri IRCCS,

Pavia (Maugeri Foundation)

Italy

Tel: + 3902527741

E-mail: [andrea.mazzanti@icsmaugeri.it](mailto:andrea.mazzanti@icsmaugeri.it)