International Journal of
Neonatal Screening

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Next Generation Sequencing (NGS) in Newborn Screening

Guest Editors:

Dr. Mei Baker
Wisconsin State Laboratory of Hygiene, Department of Pediatrics, School of Medicine and Public Health, University of Wisconsin, Madison, WI 53706, USA

Prof. Dr. Edwin Naylor
Consultant in Newborn Screening & Biochemical and Molecular Genetics, Department of Pediatrics (Genetics), Medical University of South Carolina, Charleston, SC, USA

Dr. Peter C. J. I. Schielen
Office of the International Society for Neonatal Screening, Reigerskamp 273, 3607 HP Maarssen, The Netherlands

Deadline for manuscript submissions:
closed (1 October 2021)

Message from the Guest Editors

Dear Colleagues,

In recent years, next generation sequencing (NGS) has secured a place in newborn screening. Early papers still described concepts, which especially invited a bio-ethical discussion on the pros and cons of the introduction of NGS, be it targeted or as a whole genome or exome approach. That did not stop the technical and epidemiological developments. Thus, these days, NGS technology is actually applied within newborn screening, almost exclusively in a second TIER setting, but this may change very soon. Developments concerning NGS in NBS have been quite astounding and we feel that it is time to determine where we are in this development, what is already in place, and what yet needs to be done.

This Special Issue on NGS in Newborn screening invites papers on the following:

- Application of NGS in NBS in current practice
- Proof of principle applications and retrospective studies
- Prospective pilot studies
- Application in current routine practice, both first TIER and second TIER
- Contributions concerning medical ethical issues of NGS in NBS
- QC QA issues/proficiency samples in NGS

mdpi.com/si/66750