

Special Issue

Next Generation Sequencing (NGS) in Newborn Screening

Message from the Guest Editors

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

Guest Editors

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Deadline for manuscript submissions

closed (1 October 2021)



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About the Journal

Message from the Editorial Board

The *International Journal of Neonatal Screening (IJNS)* is owned by the International Society for Neonatal Screening. The journal is a publishing platform for all topics concerning newborn screening and covers clinical presentations, metabolite pattern detection, technical aspects, and “tips and tricks”. “Tips and tricks” will appear in the form of “Technical Notes” on the journal website, thus making *IJNS* the platform for NBS laboratories to share their valuable information as to how they have improved assays, workflow, and performance. Such improvements are sometimes achieved through tiny adaptations of existing methods, which normally remain in lab journals, but which might help improve the performance of NBS laboratories around the world.

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