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Genomic Sequencing in Maternal and Child Health

Guest Editor:

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Message from the Guest Editor

Dear Colleagues,

Exome and genome sequencing (ES/GS) exert profound influence on clinical care by ushering precision medicine into routine practice. Superior to conventional genetic tests in their ability to identify disease-causing variants in certain pediatric patient populations, these technologies are gaining traction as a gold standard diagnostic. However, further evidence of their value, value for money, and ethically appropriate use as diagnostic tools in the prenatal and early postnatal period is warranted. Beyond the diagnostic application of these technologies for prenatal, early postnatal, and pediatric care, attention is turning to their application to prenatal and newborn screening, where no a priori indication for testing exists. Arguably, the recommended reporting of secondary findings identified in the context of diagnostic applications of ES/GS suggests that the use of these technologies as screening tools is already taking place. While understanding the DNA code of an apparently healthy fetus, a newborn baby, or a young child may provide invaluable guidance and optimize individual and family health outcomes, harmful consequences are also plausible.



