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CentoICU: A time sensitive targeted panel approach for genetic testing in neonatal and pediatric intensive care units

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Whole exome and whole genome sequencing allows to probe for multiple genetic conditions in a single step, reducing the time and cost to reach a diagnostic decision. These technologies have also entered the critical care units (including neonatal) where time to diagnosis is extremely important not only to guide treatment decisions and treatment efficacy but also to assess utility of procedures like surgery or transplantation. While it is not uncommon to use karyotyping, FISH, aCGH and single gene sequencing in these critical units, the emergence of NGS promises to detect both sequence variants with copy number variants within a single test, making the whole procedure significantly more effective in a clinical setting. The majority of the genetic disorders- especially autosomal recessive metabolic diseases or de novo cases- typically appear in the ICU without prior indication. Genetic testing is thus targeting a larger group of patients in the ICU where clinical features of the diseases will be more prominent than before birth or start the first time after delivery. This is particularly of importance, when one is screening for complex neuromuscular disorders or metabolic abnormalities where more than one gene can be involved. CentoICUTM offers a targeted panel based solution for early and fast diagnosis of critically ill newborns and children under 24 months. In the current format, this panel screens for several hundreds of diseases in less than 4-7 days.

Biography

Shivendra Kishore holds Bachelor's and Master's degree in Biochemical Engineering from IIT Delhi. He obtained his PhD from University of Erlangen, Germany in non-coding RNAs involved in genetic abnormalities. Post-PhD, he worked in USA, Germany and Switzerland where he further worked on alternative splicing, transcription, RNA binding proteins, small RNAs and cancer-specific recombination hot spots utilizing single gene models and also high-throughput transcriptome-wide analysis. He subsequently forayed into healthcare consulting where he worked in Basel with a focus on Market Access and Pricing. Since 2013, He heads the Department of New Technologies at Centogene that constitutes the core of R&D activities.

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